



### EDITORIAL

Because Michael Morgan is very busy these months – he has amongst other things to organize the AGM of EURO-ATAXIA of 27/28/29 September 1996 almost without assistance – I have taken over the editorship for this and the next issue of the newsletter. English is not my native language, so excuse me if my grammar or expressions aren't totally correct.

In this edition attention will be paid to the speeches held at the AGM 1995 in Lunteren by Ewout Brunt, Ria Broekgaarden and Anja Horemans. Ewout Brunt spoke at that meeting about dominant ataxias; his speech was for a major part focused on the classification into ADCA's and SCA's.

Ria Broekgaarden's subject was EAMDA, the European Alliance of Muscular Dystrophy Associations. We have put this item on the program because we would like EURO-ATAXIA to consider to somehow associate with EAMDA.

Anja Horemans explained how the VSN (the Dutch association for neuromuscular disorders) tries to improve the medical care for people with a neuromuscular disease. She also told about the way the VSN is trying to stimulate research into those diseases on a national level.

Next to this matter there is a report on a Dutch research on Friedreich's ataxia and hearing problems that is being planned at the moment.

From Cuba we received a report about the registration of and research into the dominant Cuban ataxia and the meeting organized on the first anniversary of the Cuban genetic lab.

Following on the bookreview in *Euro-Ataxia* nr.7 about eugenics and euthanasia in the first half of this century, there's another article on this ever-current question.

In the next issue we will publish an article written by Manfred Van de Kerchove and Eric Legius on predictive testing in (hereditary) ataxias. We would like to dedicate at least a major part of the next issue to this problem, so we will very much welcome other contributions on this subject.

*Carolien Koopmans*

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### HEREDOATAXIA, CLASSIFICATION AND ANTICIPATION

My talk on autosomal dominant ataxia will focus on the classification of heredoataxia, and our study of a large family in our region, with special attention to anticipation.

Due to variation in symptoms between members of the same family, and more or less overlap in manifestations of different types, the classification of hereditary ataxia has not been a straight forward task, with successive splitting and lumping. At one time, it was said that as many

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classifications existed as there were authors on the subject. It all started in 1863, when Friedreich was the first to describe the autosomal recessive hereditary type, named after him. He separated this type from the 'locomotor ataxia' which in that time was usually caused by a syphilitic infection. In 1893, Pierre-Marie drew attention to some previous reports on ataxia with autosomal dominant inheritance, late adult onset, hyperreflexia, and disturbed eye motor activity. Subsequently, different pathological types as Menzel's olivo-ponto-spinocerebellar degeneration, Dejerine-Thomas' olivo-ponto-cerebellar degeneration, and Holmes' cerebello-olivary degeneration were recognized. In the 1970's, a genetical-clinical-pathological classification was proposed by Konigsmark and Weiner, called olivo-ponto-cerebellar atrophy (OPCA) I-V. But, as was the case with previous classifications, the OPCA classification proved to be unreliable. Therefore, in 1982, Anita Harding suggested a pure clinical classification for Autosomal Dominant Cerebellar Ataxias. ADCA I-IV, to be used until the final, genetic, classification would be available. With the elucidation of the genetic characteristics, the clinical ADCA classification is now by and by replaced by the genetic 'Spino-Cerebellar Ataxia' (SCA) classification. In the current classification three groups of hereditary ataxia are recognized:

1. ataxias caused by defined metabolic diseases (usually autosomal recessive, with early onset)
2. ataxias with congenital or infantile onset
3. ataxias with juvenile or adult onset.

This group again is divided according to the mode of inheritance: autosomal recessive ataxias, like Friedreich's Ataxia or ataxia telangiectasia; autosomal dominant ataxias; and X-linked ataxias. A special subgroup of the autosomal dominant ataxias are the periodic, non progressive ataxias. One type of these periodic ataxias has been genetically identified, and is caused by a mutation in a gene on chromosome 12p, which encodes for a (cell membrane) potassium channel.

Let me summarize the ADCA and SCA classification, and their mutual relation. The largest group is ADCA I. This group contains the types of ataxia with in addition: oculomotor palsy, spasticity, or involuntary movements such as dystonia (slow, complex, writhing movements). ADCA II refers to hereditary ataxia with progressive loss of vision. ADCA III is a pure cerebellar, late adult onset type of ataxia, and ADCA IV refers to ataxia combined with myoclonus (short, simple, fast jerks) and deafness. Some of ADCA I, II and III types of ataxia now have been classified genetically as SCA1-5 and SCA7. ADCA I includes the classifications SCA1 to SCA4, ADCA II has become SCA7, and ADCA III equals SCA5. The

genetic classification has made clear that clinical manifestations may vary widely, both within and between families. However, the various SCA types do appear to have some distinctive characteristics. In SCA1 (chr6p), ataxia is often combined with spasticity and oculomotor signs as nystagmus (small jerky eye movements when looking aside). SCA2 (chr12q), also known as the 'Cuban' or 'Holguin' type of ataxia, is characterized by early occurrence of slowed saccades (jumping movements of the eyes when looking away). People with SCA3, or 'Machado-Joseph' (MJD)/ 'Azorean disease' (chr14q), largely resemble SCA1. Peculiar characteristics of SCA3/MJD are the sometimes occurring bulging eyes, weakness, loss of sensation, parkinsonism (slow movements) and involuntary movements. In SCA4 (chr16q) ataxia is combined with sensory neuropathy, very much like Friedreich's Ataxia. SCA5 (chr11c), thus far diagnosed only in the family of the American president Abraham Lincoln, is characterized by relatively pure cerebellar ataxia and late adult onset. Having mentioned SCA1 to 5 and SCA7, what about SCA6? SCA6 is still enigmatic, as this genetic type has been claimed and reserved, but has not yet been described. There is one other genetically identified autosomal dominant ataxia without a SCA number. This is a rare disease, called 'dentato-rubro-pallido-luysian atrophy' (DRPLA) (chr12p), after the affected brain nuclei. In DRPLA, ataxia occurs with myoclonus, epilepsy, and dementia. It occurs mainly in Japan and is similar to 'Haw River disease' from North Carolina. In SCA1, SCA3/MJD and DRPLA, the genes and mutations have been identified. All three mutations consist of unstable CAG repeat expansion, giving rise to abnormal proteins named respectively: ataxin 1, ataxin 3 and atrophin.

In the majority of the inherited ataxias there is no treatment. But some, caused by known metabolic disorders are potentially treatable. Treatable inherited ataxias include: selective vitamin E deficiency, Wilson's disease, Refsum's disease, neuro-acanthocytosis, mitochondrial disorders, some amino acid disorders, and the episodic ataxias. Red flags indicating a possibly treatable biochemical disease are: early onset, episodic occurrence, liver or renal disturbance, myoclonus and polyneuropathy.

Let me now describe two large families from our Groningen region, indicated as 'S' and 'B', after their principal investigators. Doctor Smit, a family physician, certainly gained a unique expertise, studying 'his' family well over 40 years. Although the primary geographical areas of the two families are only 50 kilometres apart, the pedigrees of both families are not linked at their earliest known ancestors around

AD 1800.

Discussing the features of the two families, doctor Smit and I first thought that these two families were different. As compared to the 'S' family, in the 'B' family the mean age at onset was almost 9 years later, and the progression to dependency seemed less rapid. In the 'B' family, symptoms generally were milder, with less prominent pyramidal signs (spasticity). On the other hand, other symptoms as ophthalmoplegia (paralysis of eye movements), and loss of distal reflexes occurred equally frequent in both families. During our study, two aspects became apparent, which explain these differences. First, the occurrence of certain symptoms, as spasticity, is related with the age at onset. Second, while over a length of time ataxic symptoms evolve, other symptoms may after a period be replaced, as for instance trunk (axial) ataxia which precedes limb ataxia. Similarly, there is a certain evolution of oculomotor (eye movements) symptoms. In an early stage the compensatory eye movements caused by head rotation are increased, and saccadic (jumping) eye movements are too large, showing 'overshoot'. At a later stage, when saccades become slowed, compensatory eye movements are decreased, and saccades show undershoot and splitting up. In a still later stage eye movements may be lost all together, and people have to rotate their head to look aside. Likewise early muscle tendon hyperreflexia may later change in hyporeflexia. Now the mean age at onset in the 'S' family appeared almost 5 years earlier than in the 'B' family. Therefore, the age related occurrence of symptoms, and the possible succession of symptoms may well explain the apparent clinical differences. Similar age dependent differences in clinical manifestations have been described in Machado Joseph disease. In the 'S' and 'B' families, the mean age of onset was approximately 45 years, the earliest being only 15 years, and the oldest 70 years. The age at onset in children appeared related to that of the affected parent, but could be up to 30 years earlier or twenty years later, and within one sibship the age of onset varied up to 20 years.

In 1994, we took a close look at possible 'anticipation' in the 'B' family. Anticipation means that the next generation has an earlier age at onset, and faster progression. At that time, anticipation was known to occur in some other heredoataxias, like Huntington's disease, SCA1, SCA2, and Machado Joseph disease, and in some instances anticipation occurred specially in children from affected fathers. Because of the insidious onset, we paid much attention to obtain reliable and comparable data in both living and deceased family members. Using a margin of plus/minus 4 years, anticipation was proven for the

whole family. Dividing the paternal and maternal offspring, it appeared that only children of affected fathers contributed to this outcome, with an average difference of minus 9 years.

While this clinical study was performed, the genetic type of the 'S' and 'B' families was also determined. Following a report early in 1994 by the Paris group on the SCA3 locus on chromosome 14q, Corien Verschuuren from the Groningen department of medical genetics, proved linkage to the SCA3 locus in both the 'S' and 'B' families. In addition, from the similarity of the haplotype (variety) of the genetic markers around the SCA3 locus, it appeared that the 'S' and 'B' families were linked, and part of one larger family. This again proved that the apparent clinical differences were indeed manifestations of the same disease. Now, this SCA3 locus was actually within a larger region, to which Machado Joseph disease had been linked before, but with apparent clinical differences, the Paris group judged SCA3 and MJD to be genetically separate. In november 1994, Japanese researchers on Machado Joseph disease, described the gene itself with the mutation, another CAG repeat extension. After this report, we looked at this mutation in 'our' families, and found the same mutation, proving that SCA3 and MJD were genetically identical. As had been found in SCA1, in our large family, an earlier age of onset was related to a larger expansion of CAG repeats. However for any given number of CAG repeat a wide variation of up to 35 years existed. Thus, the number of CAG repeats did not provide an individual prediction of age at onset. Comparing the numbers of CAG repeats in paternal and maternal offspring, we found a further CAG expansion in most paternal but not maternal transmission. Combining the clinical and genetic data, might a CAG expansion explain for the anticipation? It appeared that a further CAG expansion could only partially explain the occurrence of anticipation, so we still have to find other factors. Meanwhile, other groups have also demonstrated anticipation in SCA3/MJD, and anticipation has now been reported in all SCA's.

What made the research into ataxias especially exciting to me, is that we may be getting at the understanding of a specific neuronal degeneration, and this may very well reveal more general mechanisms, which apply to other degenerative diseases such as Alzheimer and Parkinson's disease. My special thanks go to Anneke Joosten, who did the marvellous work on anticipation in this family.

*Ewout Brunt*

## EAMDA

Thank you for your invitation.

I would like to introduce myself: I am Ria Broekgaarden, I am employed by the VSN (Vereniging Spierziekten Nederland). VSN is a member of EAMDA (European Association of Muscular Dystrophy Associations), and through VSN I am involved with EAMDA and more specific with Eastern European affairs.

The VSN is the Dutch umbrella organization of neuromuscular diseases (NMD). One of the NMD for which the VSN caters is Friedreich's ataxia.

By the way, both VSN and EAMDA started as organizations for muscular dystrophy, but have expanded their activities to other muscular diseases and at the moment are devoted to neuromuscular diseases.

At this moment the AGM of EAMDA is in session, in a conference center not very far away from this one. One of the subjects at hand within the board is the participation in, membership of and cooperation with diagnosis bound groups. In this respect Hans Doré presented your organization to the participants of EAMDA yesterday.

**What is EAMDA?** EAMDA is an European alliance of independent national associations for NMD. Its activities are carried out primarily by the member organizations that offer to work out certain jobs. The national groups may also contribute to what other member organizations are planning to do.

The strength of EAMDA is that it is founded on consumer-based national associations, each of them trying to achieve improvement of conditions of life for people with NMD and giving support to research into these diseases. This is done with the help of professionals but it is the people with NMD themselves who formulate their wishes.

**Achievements.** EAMDA wants to exchange ideas and to discuss policies between its members. In this way EAMDA tries to increase the level of activity within the alliance as well as to develop mutual cooperation among the members for all to work with greater efficiency and with a broader range of projects.

EAMDA supports the NMD organization in Eastern Europe.

EAMDA also started a worldwide network (WAMDA) and European research network ENMC (European Neuro Muscular Centre).

**Members.** 30 Countries from all over Europe are at this moment member of EAMDA: Austria, Belgium,

Bulgaria, Croatia, Cyprus, the Czech Republic, Denmark, Estonia, Finland, France, Germany, Ireland, Italy, Lithuania, Malta, Republic of Moldova, the Netherlands, Norway, Poland, Portugal, Romania, Russia, Slovakia, Slovenia, Spain, Sweden (2x), Switzerland (2x), Turkey, United Kingdom, Yugoslavia.

**Neuromuscular diseases.** Neuromuscular diseases is a collective name for a number of diseases in which the nerves and/or muscles are affected. Most of the diseases are hereditary and progressive. Some are congenital. Many people with NMD will need a wheelchair someday and become increasingly dependant on care. There is yet no cure for most neuromuscular diseases. However, it is often possible to maintain some skills and to delay – by means of therapeutic treatments – some of the consequences of the diseases, for instance stiff joints, scoliosis (spinal curvature) and respiratory problems. There are a lot of NMD (about 600).

Not all member organizations of EAMDA are involved with ataxias. Some of them only deal with SMA and Duchenne dystrophy.

**Objectives of EAMDA.** The objectives for which the alliance is established are:

- to improve the quality of life for people with NMD;
- to promote research into all aspects of NMD and to assist such research whenever appropriate;
- to provide a network of NMD-associations across Europe with the primary purpose of 1. giving support, assistance and advice and 2. facilitating an exchange of information on topics which are of mutual interest to such associations.

To realize these objectives EAMDA will have the following powers:

- to arrange meetings and promote workshops which will disseminate information on developments, treatment and therapy concerning NMD and to publish the results;
- to develop and improve research in the disciplines connected with NMD and with early diagnosis, treatments and rehabilitation services in that field;
- to coordinate and develop the means for the collection, distribution and exchange of specialized knowledge;
- to collect and disseminate information on NMD in general.

The goals that EAMDA is dedicated to are:

- to improve quality of life for people with NMD;
- to promote research into NMD;
- to provide a network of NMD-associations;
- to arrange meetings and organize workshops;
- to develop and improve research;

- to exchange specialized knowledge;
- to collect and disseminate information.

**Cooperation.** The main objectives of EAMDA are to provide a network of NMD associations across Europe with the primary purpose of giving support, assistance and advice; facilitating an exchange of information on topics which are of mutual interest to such associations and to improve the quality of life for people affected by muscular dystrophy or other neuromuscular disease.

**Benefits of cooperation.** The common interests and goals, resulting in similar organizations and activities, leads to more efficiency and effectiveness. Because we have goals in common (for example to promote research) and have activities in common (e.g. internet), we can be more efficient and effective.

**The disadvantages of cooperation.** Although it can be more effective, more efficient to work together, it also can delay and make it more difficult to reach a certain goal for a specific group.

You, my audience, are representatives of national organizations cooperating on a European level. That is why I have informed you of EAMDA and not of the VSN. Thank you for your attention.

*Ria Broekgaarden*

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## RESEARCH INTO A UNIFORM PROTOCOL OF CARE FOR REHABILITATION DOCTORS

I would like to speak about a project which my organization (VSN, Dutch Association for Neuromuscular Disorders) has started in collaboration with rehabilitation doctors. The VSN is an umbrella association for people with neuromuscular disorders (NMD). It has 13 diagnosis bound groups and the Friedreich's Ataxia group is one of them.

The goal of the project is the realization of a protocol (guideline) of care for rehabilitation doctors. To be able to understand the protocol and the need for it, you have to know how rehabilitation in the Netherlands is organized.

Rehabilitation is organized differently from country to country. In the Netherlands there are different kinds of rehabilitation facilities. We have 700 to 800 hospitals and each of them has its own department of rehabilitation. Most people with NMD and also most people with ataxia will not visit such a department often. Only when you recover from an operation, rehabilitation will start in this department. In other situations care is provided by one of the 40 rehabilitation centres in the Netherlands. Especially persons with NMD and those with ataxia receive

treatment from these centres.

There is another type of rehabilitation in the Netherlands which is provided by therapists who live in the direct environment of the 'patient': the physiotherapist. In the Netherlands there are 5 to 10.000 physiotherapists. Many persons with FA receive rehabilitation from such a therapist. Although it may be useful to get help from a local therapist, the major drawback is the little experience with FA of these physiotherapists. Therefore the VSN advises people to go to rehabilitation centres. In these centres the care is provided by a multidisciplinary team, coordinated by a rehabilitation doctor. Rehabilitation doctors are very well organized in the Netherlands and intensive contacts exist with the VSN.

The above mentioned protocol is mainly focused on the rehabilitation doctors employed in these centres and will be formulated with the help of some of these doctors.

Rehabilitation is important for the VSN members. No other medics see these patients so often on a regular basis. The VSN wants to improve the quality of the care, but for this the support of the care providers in rehabilitation centres is necessary. Together with these providers the VSN has started several activities to improve the quality of care. For 20 years the VSN has been initiating contacts between rehabilitation doctors and therapists. As a result there are now about 50 doctors in the Netherlands who are 'experts' in the rehabilitation care of people with NMD. Once a year the VSN organizes a symposium for doctors and therapists. In addition to these activities the VSN is active in stimulating researchers to study these rare NMD.

The goal of the project is to instore a protocol (a kind of guideline) of care for people with NMD. This protocol can also be used by other doctors. Up to now doctors plan a treatment for each patient only based on their own experience and without specific literature support. This results in different treatments for the same disorder. In a uniform protocol each patient undergoes the same examinations and gets the same treatment. For example: people with FA (especially those above a certain age) are advised to consult a cardiologist regularly (maybe once or twice a year). With the protocol rehabilitation doctors, therapist and even you, will know which examination is necessary and which is the most suitable treatment.

Currently the VSN is developing a similar protocol for the neuromuscular disorder HMSN (Hereditary Motor and Sensory Neuropathy), in collaboration with 8 rehabilitation doctors. The experience obtained by this project may be of help in developing guidelines for other diseases in the future.

Each neuromuscular disorder needs his own protocol because each disease has specific symptoms. We hope to develop a protocol for FA in the future.

*Dr. Anja Horemans*

## FOR DREAMS NOT TO BE FRUSTRATED

Ataxia just arrived to my family with the disbalance in gait and the dysarthria of my paternal grandmother. It was something without name, inherited from my great-grandfather, being an unknown disease at that time. Nobody thought about what it would mean for our future life.

Later on, my uncle got ill when he was thirty-eight years old. But it was not until my dad began to show his dullness in his fifth decade of life, that those experiences began to be traumatic for my family.

In the year 1983 the neurologist G. Orozco was making the first steps in the study of that progressive disease that invalidated a great number of inhabitants of Holguin. Holguin is the province of Cuba that is distinguished from the other provinces of our country by the high occurrence of ataxia disease.

And it was Dr. Orozco himself who answered a lot of questions accumulated during many years. He made us understand the irreversible reality of being genetically marked by the disease named Cuban ataxia. Studying that disease has been a scientific medical challenge which Dr. Orozco took on personally.

More than ten years passed and many expectations are reality now already. The silence regarding the disease has been broken. Dr. Orozco has consolidated his research team, and is leading a Cuban-German project on the island. This team, small but very well organized, has got the first Cuban molecular genetic lab for ataxia studies.

In this small lab samples and data from all affected families have been collected in a preventive research program having three main objectives: 1. to create a DNA bank; 2. to make a presymptomatic diagnosis possible; 3. to make a prenatal diagnosis possible. The program is aimed at guaranteeing healthy descendants in the coming generations.

Thousands of Cuban inhabitants who are now suffering from ataxia as well as their relatives still keep their hopes on this German-Cuban research project led by Dr. Auburger and Dr. Orozco.

Only one year after the creation of the Cuban lab many achievements have been obtained, such as the realization of a DNA bank, the creation of a computerized genetics register of all affected families, as well as the development of a supporting team composed of a psychologist, a psychiatrist and genetic specialists for presymptomatic diagnoses.

On the first anniversary of the genetic lab a meeting of researchers, people having to do with public health and state representatives in the province, as well as patients suffering from ataxia and their relatives was organized. Mr. Felipe Pena, specialist in sport and suffering of ataxia, spoke the following words on this occasion:

“I am the son of an ataxic person and I also am affected by the disease. I know my destiny will be

a wheelchair and a bed, but I have got big hopes on that lab, due to the fact that with that lab my sons will already know whether they are going to get sick or not. If they do not get sick they may live a normal life and could reach goals as normal human beings do. If the opposite occurs Doctors will know how to prepare them to carry on their ailment through life. This lab is the opening of a very difficult road aimed at getting a solution for our lives. I would like to express my gratitude here and give thanks to our Government, Dr. Auburger, Dr. Orozco and their team, and to all persons who struggle to improve our existence.”

Mr. Ramon Diaz Alcantara and Mr. Meinardo Friman congratulated the research team for their achievements, and they stated that this team will always have the support of the government.

At that emotional moment I felt obliged to express my gratitude in front of those who have dedicated part of their lives to improve our life. I thanked them on behalf of more than five hundred sick persons and thousands of relatives in our province.

This lab now is a dream come true which will put an end once and forever to the uncertainty of one part of the population likely to have sick sons. And sometimes this fact forced most of us to accept the idea to be under an unavoidable natality control.

Soon afterwards Dr. Orozco spoke about yesterday's efforts, today's results and their hopes for tomorrow and the will they have to continue with their research. While Dr. Orozco was talking I thought about my father who is fifty-eight years old now, who has been prostrated during twelve years and now his critical condition does not allow him to rejoice to the hopes of this group of researchers.

I also thought about thousands of human beings in the world who have not lost their hopes. And I wished deeply that the hopes of all these scientists who struggle for a better future giving the best of their own for their dreams not to be frustrated as well as the big dreams of my little son and many others who were born innocent but have the chance to be affected by this diabolical genetic disease.

*Hilda Pupo Salazar*

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## EUGENICS AND EUTHANASIA

*Slip slidin' away*

*Slip slidin' away*

*You know the nearer your destination*

*The more you're slip slidin' away*

The subject of Michael Morgan's bookreview in *Euro-Ataxia* nr.7 was indeed very interesting. I must confess that I haven't read *Death and Deliverance* myself, but because I have read a lot about history and because the bookreview was so well written, I had no problem following Burleigh's line of reasoning.

Michael and I think the subject is important enough to philosophize on a bit more.

My starting-point is the following. Evolution has equipped homo sapiens with a consciousness. According to Darwin this consciousness must be a means through which the chance of survival of human beings can be heightened. In my opinion the human consciousness can indeed make it easier for man to survive. Therefore man should keep one condition in mind: there should always be a balance between his intellectual capacities and his feeling, i.e. the analyzing and the integrating part of his thoughts.

Now the entire world was out of balance for a long time already (for centuries or maybe millennia) at the end of World War I. The Treaty of Versailles, the peace-settlement agreed on after the First World War, only added to this imbalance. Europe and the USA put the blame for that war on Germany and demanded heavy Reparation Payments from it. This was one of the circumstances under which the German economy collapsed. A high level of unemployment and poverty were the result, with people losing all hope of ever finding a job again. There was an enormous feeling of discontent throughout entire Germany. And then came Hitler, who offered some kind of solution. By starting to make preparations for war, the economy flourished again. The people in Germany just closed their eyes and hearts for the bad sides of the nazi-system and profited from the good sides. To use 'my' terms: there was no balance between feeling and intellect, but most Germans didn't care to give the nazi-ideology a second thought. As the Beatles sing in *Strawberry Fields Forever*: 'Living is easy with eyes closed, misunderstanding all you see'. History is full of examples of that hypocritical behaviour. It is not a conduct that was only shown during the nazi-period by the Germans. The consequences simply never were so extreme as during the Third Reich.

The Eugenics program, just like all things in nazi-Germany, could happen because the mental or moral balance was completely neglected. Mengele c.s. were welcomed to do terrible genetic experiments with living human beings – with a preference for experiments on Jewish twins. Under the pretext of enlarging their scientific knowledge, the nazi-doctors could be as cruel as they liked, satisfying their curiosity or just for fun. The slaughter of a mass of *Untermenschen* was another means to reach a purification of the so-called Arian race. The computer-like precision and the lack of feeling with which the Eugenics program was carried out, was appalling.

Let's return to Darwin. In the animal world all species have feedback mechanisms which adjust the rate of breeding so as to keep the population at a stable level. In recent years biologists have discovered that every animal species which they studied – from flower beetles through rabbits to baboons – is equipped with instinctive behaviour-

patterns which put a brake on excessive breeding, and keep the population-density in a given territory fairly constant, even when food is plentiful. When the density exceeds a certain limit, crowding produces stress-symptoms which affect the hormonal balance; rabbits and deer begin to die off from 'adrenal stress' without any sign of epidemic disease; the females of rats stop caring for their young, which perish, and abnormal sexual behaviour makes its appearance.' (A. Koestler, *The ghost in the machine*, London 1967, p. 329). Looking at the human overpopulation, you could say that the broader European eugenic movement tangled a real problem. The fact that people misused the eugenic ideas as a rational argument for their egoistic feelings of fear and distress, doesn't mean that the eugenic ideas are senseless or criminal. People who are right for the wrong reasons – and vice versa – are all around.

The discussion about euthanasia is troubled by the same moral questions as the eugenic movement. After much discussion the Dutch euthanasia-law finally passed Parliament. The reactions from outside our country are sometimes very misunderstanding, so let me explain the Dutch policy regarding euthanasia.

In the Netherlands euthanasia is still a crime according to the Penal Code. But under some very strict conditions the Public Prosecutor will not prosecute cases of euthanasia. The euthanasia has to be a request from the patient himself. It must be a case of severe mental or physical suffering for which there is no cure. The doctor of the patient has to ask the advice and permission of a colleague. Only a doctor can perform euthanasia, not a nurse or a husband or wife or someone else. And the euthanasia has to be reported to the Public Prosecutor. If he thinks that the euthanasia is performed not according to these strict rules, he has to refer the case to the courts.

The motive for the Dutch euthanasia-law is the respect for life, not the desire to get rid of nasty, worthless and expensive handicapped and old people. We in the Netherlands are convinced that you can't express the value of a human life, certainly can't count it in money. Sometimes a patient can have a disease for which there is no cure and the only thing that he can do is lay in bed with a lot of pain and wait for death. If the unescapable death will come with even more pain or by suffocating slowly, why shouldn't we be so humane to let that patient die quietly and peacefully? Why should such a patient go all the way and suffer intensely? In the Netherlands we think it is in some cases morally allowed to give such a patient an injection which will send him to sleep and induces the heart to stop beating after a while. But the cases in which euthanasia will not be prosecuted have to respond to severe criteria. Because only a person himself can judge the value of his life, a doctor who has performed euthanasia is not prosecuted *only* if the patient himself has requested for it. The doctor furthermore is not allowed to judge

on the sincerity of the patient's wish to die on his own, but always has to consult a second doctor. And all cases of euthanasia have to be reported officially. I am proud that the Dutch dare to bring the issue of life and death out in the open. When one takes a position in this matter, one risks to be called immoral. The pope for instance declared that the Dutch euthanasia-law was the legalization of murder and said that the Dutch government was a bunch of criminals. He says that we should leave the decision about life and death in the hands of God, but don't we take that decision out of his hands all the time? Medical science has now reached a level at which it is possible to keep people alive who otherwise would die. Everytime we give someone a medical treatment to prevent him from dying, we in fact take the decision on life and death in our own hands. Nobody – not even the pope – complains about that reality.

There is still another inconsequence regarding the matter of euthanasia which you could indeed call hypocritical. The beautiful social security system in our country, by which the position of the weaker ones in society was protected, is being broken down at a rapid speed at the moment. Under the pretext of having to be compatible with the rest of Europe. The people who argue that euthanasia is immoral are at the same time the people who want to diminish the social security expenses. Pretending to care about the handicapped and elder people so much that no-one may let them die, but not caring enough for them to let them live a life that's worthwhile. In Holland there are nowadays a lot of people who ease their own conscience by telling themselves that they are so broad-minded and sensitive that they respect all human beings, but can believe this themselves only as long as they are not confronted by the handicapped and old people daily. We call this the 'Back behind the windowpanes'-idea. In my opinion this has nothing to do with respect for life, which they claim it has.

But the path we have chosen in the Netherlands is a slippery slope indeed. We have to be careful that any case in which the strict conditions aren't paid respect to will be prosecuted. I want to end this article with a few lines from the same song with which I opened. The lines are from a song by Paul Simon (from Simon & Garfunkel) *Slip slidin' away*. The beauty of those lines lies in the fact that they may apply to the pope as well as to Dutch government. Who knows?

*God only knows*

*God makes his plan*

*The information's unavailable*

*To the mortal man*

*We believe we're gliding down the highway*

*When in fact we're slip slidin' away.*

*Carolien Koopmans*

## RESEARCH INTO FA HEARING PROBLEMS

Like a lot of FA'ers I'm having hearing problems. When I am in a quiet room and talking with one other person or a group of people with everyone waiting for his turn to speak, I have little problems. But when I am in a noisy surrounding with a lot of people talking at the same time, I can't understand anything. All sounds seem to be alike in loudness and I can't distinguish one voice from the other. To communicate with somebody is impossible for me in crowded places.

For years already I am complaining to the doctors about my bad hearing. I did some hearing tests that only showed there was little actual loss of hearing. They nevertheless let me try some hearing aids, when I asked for it. The hearing aids were of no use because they amplify the background noise as well as the voice you want to hear. When you use such a hearing aid in a crowded room you go crazy of all the loud sounds bombarding your ears.

But because I kept complaining and the last doctor I was visiting was getting tired of my nagging, I was sent to an audiological centre (a clinical centre specialized in hearing problems). There I had to undergo some speech-and-hearing tests and a brainstem test. A short time after the tests I had to consult Jan Feenstra, the audiologist.

What a nice surprise the visit to Jan Feensta was! Not only did he know about FA and its hearing problems, but he also was the first one who said that my case was not hopeless. Whereas most people can distinguish sounds that are 5 decibel softer than the background noise, the tests pointed out that I could only distinguish sounds that were 6 decibels louder than the background noise. That makes a difference of 11 db. And that's a lot! In my case the quality of sound had to be improved, not the quantity (i.e. the loudness of sound). The quality of sound is improved when the sound reaches the ear in a more direct way, without the interference of background noise.

Jan Feenstra lent me the Hearit, a headphone connected with a microphone. When I have difficulty understanding somebody, I ask him to speak into the microphone and that really improves the communication. At small disciplined meetings – meetings at which everyone waits for his turn to speak – I put the microphone on the table and can follow the conversation without much problems.

A great advantage of the Hearit is that with the microphone on the table it reflects my own voice too. So I can better hear myself speaking. That feedback gives me more self-confidence and therefore I also speak more easily myself. The other advantage of the Hearit is that I feel myself more part of the surrounding group, more awake. In biological terms it could be expressed in the following way: my arousal-level is heightened thanks to the improved offer of auditory signals.

But the Hearit isn't always an improvement. The

acoustics of the room you're in remain of major importance. When you're in a hollow room with much resonance and with a lot of people, you still go crazy of the overwhelming mass of noise. And a mumbler or inarticulate speaker is still hard to understand. So the Hearit does help, but not in all situations.

Jan Feenstra had never met a FA'er till last year when he was consulted by three of them. All three had the same problems and the same experiences with doctors. When he looked in the scientific literature to see what was written about FA and hearing problems, he couldn't find almost anything. Only some vague terms like 'deafness' and 'bad hearing'. The percentage of FA'ers having hearing problems was somewhere estimated to be 10 to 20%. In my opinion that is far to low an estimate. Most FA'ers I know developed hearing problems sooner or later. A subject that deserves some research.

Jan Feenstra suggested to do some research himself. One doesn't have to make such an offer to the VSN twice. So at the moment Jan Feenstra, Anja Horemans (the person at the VSN-office responsible for the stimulation and coordination of scientific research), Wim Nas (the biologist of the FA-group), Hans Doré and I are developing a research-project. In a following issue of *Euro-Ataxia* Jan Feenstra will write an article about the resulting research.

*Carolien Koopmans*

## ATAXIA.NET

At this moment we can announce that in the near future EURO-ATAXIA will have its own World Wide Web-site. Thanks to VSN in the Netherlands who are kind enough to give us some space on their server. At this moment we cannot provide you with an address yet. But if you check regularly the VSN-site (<http://www.vsn.nl>) you will find a link to EURO-ATAXIA as soon as the EURO-ATAXIA pages are established.

But even without EURO-ATAXIA on the Web there is enough to find. For this issue we gathered some interesting links.

### Associations

#### National Ataxia Foundation (USA)

<http://www.ataxia.org>

#### Muscular Dystrophy Association (Australia)

<http://www.mda.org.au>

#### Vereniging Spierziekten Nederland (Netherlands)

<http://www.vsn.nl>

#### Vlaamse Liga voor Ataxie van Friedreich en andere erfelijke ataxieën (Belgium)

<http://www.club.innet.be/~pub00028/vlaf.htm> (Dutch)

<http://www.club.innet.be/~pub00028/vlafe.htm> (English)

#### ADCA-Vereniging Nederland (Netherlands)

<http://www.spin.nl/adca0301.htm>

### Disability Resources

#### Handicap Info

<http://www.globalxs.nl/home/h/hinfo> (Dutch)

<http://www.globalxs.nl/home/h/hinfo/uk/indexuk.htm> (English)

#### Disability Related Resources on the Internet

<http://disserv.stu.umn.edu/other.html>

#### Disability Resources from Evan Kemp Associates

<http://disability.com>

### Neurological and medical Information

#### OMIM - Online Mendelian Inheritance in Man

<http://www3.ncbi.nlm.nih.gov/omim>

#### Neuromuscular & CNS Syndromes

<http://neuro.wustl.edu/neuromuscular/syaltbrain.html>

**CLOSING DATE FOR  
THE NEXT ISSUE**

**1 OCTOBER 1996**



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